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Prenatal diagnosis of fetal acrania using two and three dimensional ultrasound

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Abstract

Malformations of the central nervous system are one of the most common congenital anomalies of the fetus. In this report we aimed to summarize a case of fetal acrania diagnosed in the first trimester and the use of two and three-dimensional ultrasound in early diagnosis of the disease.

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Introduction

Central nervous system (CNS) malformations are the second most frequent category of congenital anomalies, after congenital heart diseases. Most of the time ultrasound (US) examination is effective for the detection of these anomalies even in the first trimester of the pregnancy. Exencephaly or acrania is the absence of the entire or a major part of the fetal calvarium. Brain tissue is present but is not protected by a bony sheet. Sonographic assessment of the fetal skull contour in the first trimester in axial and sagittal planes and measurement of the bi-parietal diameter almost always ensure early diagnosis of exencephaly. With the progression in fetal imaging technology three-dimensional (3D) US has become an important tool for fetal

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assessment. After the introduction of 3D US for fetal imaging additional benefits for the diagnosis of congenital CNS anomalies has been shown in many studies.[1,2]

Figure I. Irregular, non-ossified fetal head

Case Report

A 20 year old primigravid woman was referred to our clinic at the 11th gestational week of pregnancy for first trimester screening. Medical anamnesis was uneventful. She was taking 400-µg/day folic acid regularly. Two-dimensional (2D) US (Voluson 730 Pro, GE Healthcare, Milwaukee, WI) exam revealed a live, singleton fetus at the 11th gestational week. The fetal calvarium was irregular in shape. There was no ossified bony structure at the upper part of fetal head and brain tissue was exposed to amniotic fluid and floating in it (Figure I). Cerebral hemispheres were present and falx cerebri was visible (Figure II). Three-dimensional US revealed an abnormal cranium. The fetal brain was present but it was not protected by bony structures; the outer shape of the head is bi-lobed. The fetal face was irregular and fetal eyes were prominent. The nasal bone was present. There were no other malformations noted on US examination. Fetal magnetic resonance imaging was not diagnostic.

The family was informed about the condition and the pregnancy was terminated with medically induced abortion (misoprostol). Gross examination of the fetus confirmed the diagnosis of exencephaly (Figure IV). There were no other major anomalies. Pathologic examination showed a lack of bony structures at the upper part of fetal head and brain tissue was covered with a thin layer of endothelium.

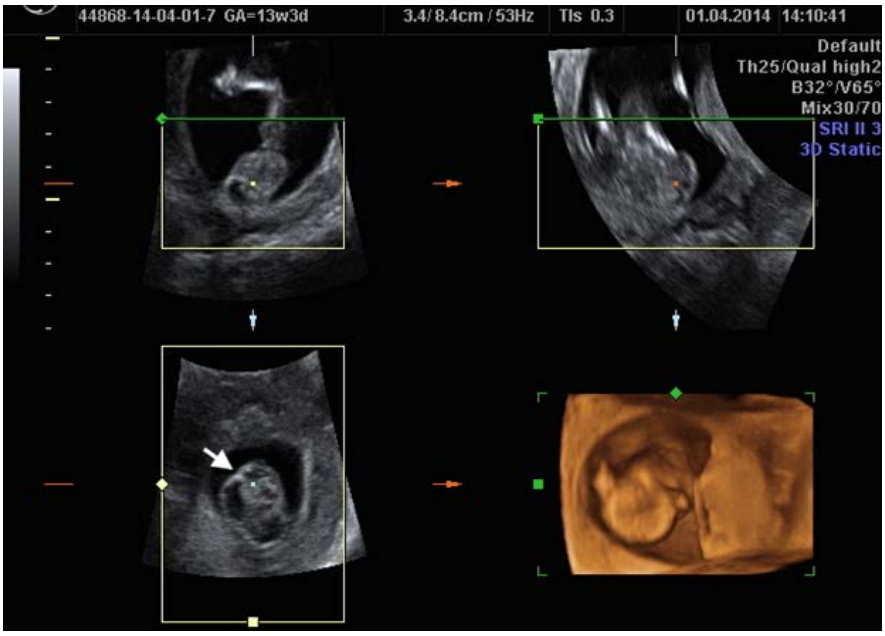

Figure II. White arrow: Falx cerebri

Figure III. White arrow: Abnormal fetal head on 3D US

Figure IV. Absence of the upper part of the bony head

Discussion

Central nervous system anomalies are the second most common fetal anomalies, after cardiac anomalies (1/100). Anomalies of the neural tube are multifactorial in origin. Chemicals, maternal status (DM), environmental factors (obesity, hyperthermia), therapeutic agents (valproate), deficiency of nutrients (folate), genetic syndromes (Meckel Gruber, trisomy 13) may increase the incidence of neural tube defects.[3] Exencephaly refers to the absence of the entire or a significant portion of the calvarial bones, but brain tissue is present. It is a rare anomaly resulting from failure of the mesenchyme to migrate under the ectoderm overlying the brain tissue to form the bony tissue over the cerebral hemispheres.[4] The risk of recurrence for neural tube defects (NTD) is approximately 2-4% with one affected sibling. With two affected siblings, the risk is approximately 10 %.[5]

The cerebral hemispheres are anatomically disorganized. Acrania is the most common CNS anomaly detectable in the first trimester.[6] Exencephaly usually progresses to anencephaly in the followings weeks of gestation. It must be kept in mind that the calvarial bones are not fully calcified before 10-11 weeks; therefore, first trimester diagnosis must be made with caution.[7] Ultrasound is the main tool for diagnosis. Classical findings in 2D US exam are:

1. Lack of cranial bones or calcification at the upper part of fetal head
2. The surface of the cranial pole appears abnormal
3. Deformed and dorsally bulging abnormal head.

With the improvement in fetal imaging technology 3D-US became an important tool for the assessment of fetal anatomy. Additional gain of 3D-US to detect fetal CNS anomalies has been reported in the literature.[2,8] Three-dimensional ultrasound has facilitated accurate and objective diagnoses of cranium bifidum/spina bifida, holoprosencephaly and associated anomalies in the first trimester and may allow detection of pathologic central nervous system (CNS) development at an earlier gestational age.[9] It is important to examine fetal head development in all planes since it will be skipped in sagittal plane. Three-

dimensional US shows prominent inter-hemispheric cerebral fissure and dorsally bulging brain (Mickey Mouse Head) (Figure III).[5,10,11]

Three-dimensional US allows analysis of images off-line as well as measuring diameters and volumes that may increase diagnostic accuracy.[12] The ability to assess volumetric structure of the brain or other parts of the fetal head gives us new clues about diseases and the opportunity of designating characteristics of congenital malformations. Surface and volume rendered images provided by 3D-US help to present the exact level of problem. Also images obtained with 3D-US make it easier to understand the extent of the disease for both the physicians and the patients.[13] The ability to rotate images is also useful in the evaluation of some of the CNS anomalies.[1] New technologies such as HDlive image rendering may be very useful for the determination of structural anomalies of the developing fetus.[13,14]

Conclusion

Acrania is a severe and lethal malformation of the CNS that can be detected in the first trimester. Three-dimensional US may help to clarify the extent and severity of the disease both for physicians and patients.

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